Some important points to remember about

Prenatal Testing

- Prenatal testing can give couples the best possible chance to have a healthy baby.
- About 95% of the women who have prenatal testing get good news from the laboratory.
- Anyone who is considering a specific prenatal test (or any other medical procedure) should learn all she can about the procedure itself, its purpose, its risks, and its benefits. She should also learn about the alternatives that are available to her and about any future decisions she may be to make based on the test results.
- Many genetic conditions can be detected and diagnosed prenatally. However, there are genetic conditions that cannot be diagnosed prenatally.
- Though no genetic condition can be cured, doctors can treat some of the effects of some genetic conditions. However, a couple who wants prenatal testing should prepare themselves to learn that their baby could be born with an untreatable condition.
- Knowing about a genetic condition ahead of time can give a couple time to prepare emotionally, financially, and in other ways to care for a special child. It can also prepare doctors to be ready with treatment as soon as the child is born. Selectively terminating an affected fetus is another option when a couple learns of a fetus' condition before it is born.

Screening Tests:

Screening tests can only tell if a fetus is at a higher risk than most for having a condition. They cannot diagnose the condition.

- **Maternal Serum AFP/HCG/UE3** (also called **Triple Screen**) is a screening test any woman can have when she is 15 - 20 weeks pregnant. This blood test measures the levels of *alpha fetoprotein, human chorionic gonadotrophin*, and *unconjugated estriol* in her blood. Alpha fetoprotein is a protein the fetus makes. Human chorionic gonadotrophin and unconjugated estriol are hormones the placenta produces.

The test can detect 85% of the fetuses that have open neural tube defects like spina bifida. It can also detect about 65% of the fetuses that have Down syndrome and some of the fetuses with trisomy 18.

- **Maternal Serum AFP** is a screening test any woman can have when she is 15 - 20 weeks pregnant. This blood test measures the level of *alpha fetoprotein* in her blood.

The AFP test was the only screening test of its kind for many years. Now, however, the Maternal Serum AFP/HCG/UE3 test has made the AFP test somewhat obsolete. The AFP test can detect 85% of the fetuses that have open neural tube defects like spina bifida, but only detects Down syndrome 25% of the time. It may still be the best screening test for some pregnant women, especially those who are too far along in their pregnancy for the Triple Screen test.

Diagnostic Tests:

Diagnostic tests can tell for sure if a fetus has a specific condition or not.

- A **sonogram** (also called **ultrasound scan**) can detect major physical defects that can be seen early in a pregnancy. During a sonogram, an ultrasound wand placed on a pregnant women's abdomen sends sound waves into the womb. The sound waves bounce off the fetus and project an image onto a screen. The doctor can then actually look at the fetus and see how it is developing. The sonogram exam poses no known risk to the fetus.
- **Amniocentesis** is a diagnostic test that makes it possible to look at a fetus' chromosomes and see if there are too many or too few of them and if they look like they should. During amniocentesis, a doctor takes out a few tablespoons of amniotic fluid through a special needle he puts through the woman's abdomen into her uterus. The fetus' chromosomes are present in the fetal cells in this fluid. A woman who wants amniocentesis should usually have it between her 14th and 16th week of pregnancy.

If doctors think a fetus may have a condition that is caused by a specific gene, amniocentesis can sometimes diagnose that condition. An example of this kind of *single-gene disorder* would be cystic fibrosis. Amniocentesis can also detect open neural tube defects like spina bifida.

Amniocentesis isn't for every pregnant woman. Only a woman who is 34 or older or whose family members have an inheritable condition that amniocentesis can detect should have the test. The procedure can cause a miscarriage or other problems. The chance of it causing a problem is very slim (½% or less), but it does exist. Before a woman has amniocentesis, the fetus' chance of having a genetic condition should be greater than the test's chance of causing a problem.

Genetic Screening & Case Management
Bureau of Children's Health • Texas Department of Health
1100 West 49th Street • Austin, Texas 78756-3199
512-458-7700